Figure 1

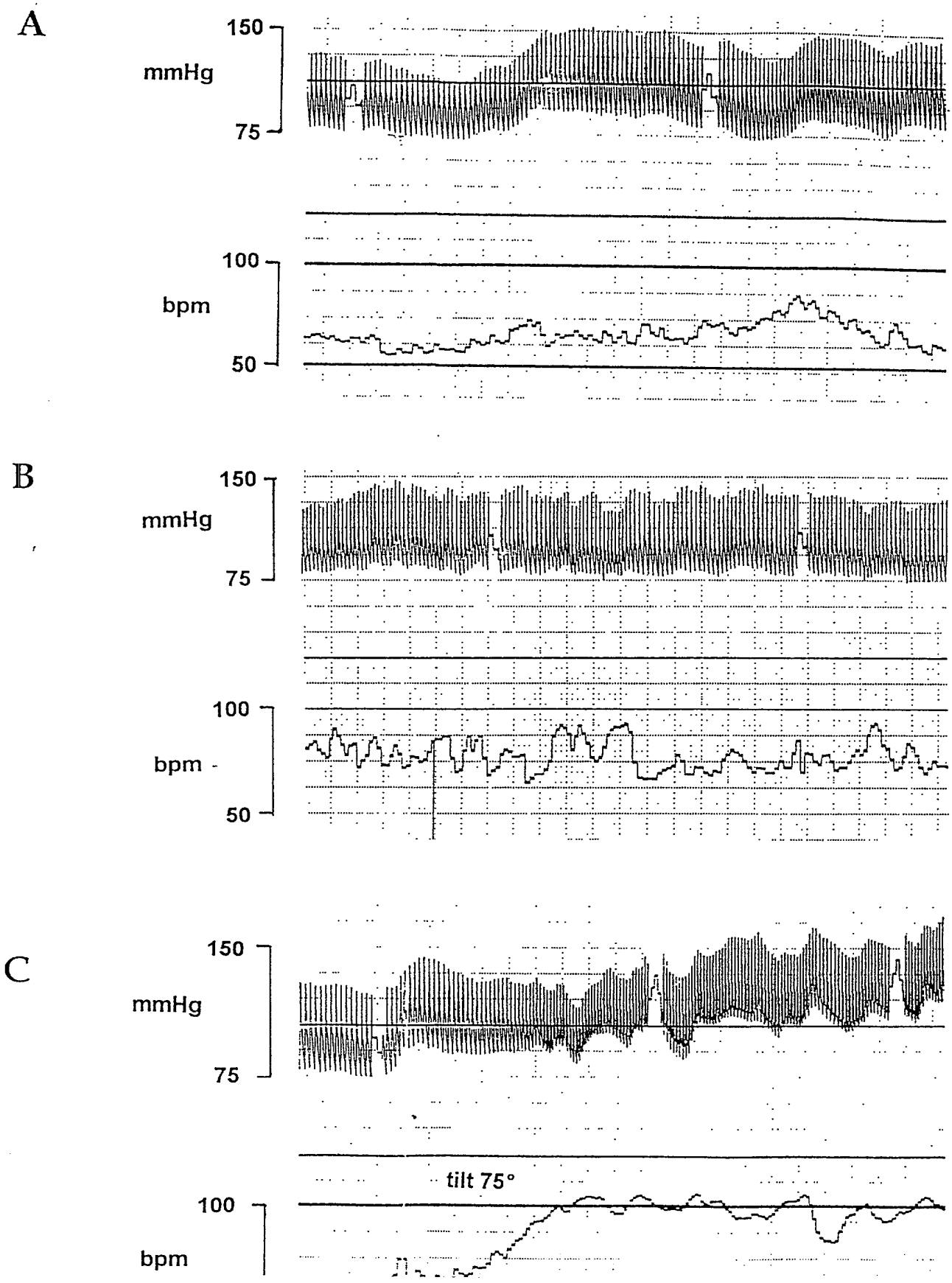

A
150
mmHg
75
100
bpm
50
B
150
mmHg
75
100
bpm
50
C
150
mmHg
75
tilt 75°
100
bpm

Figure 2

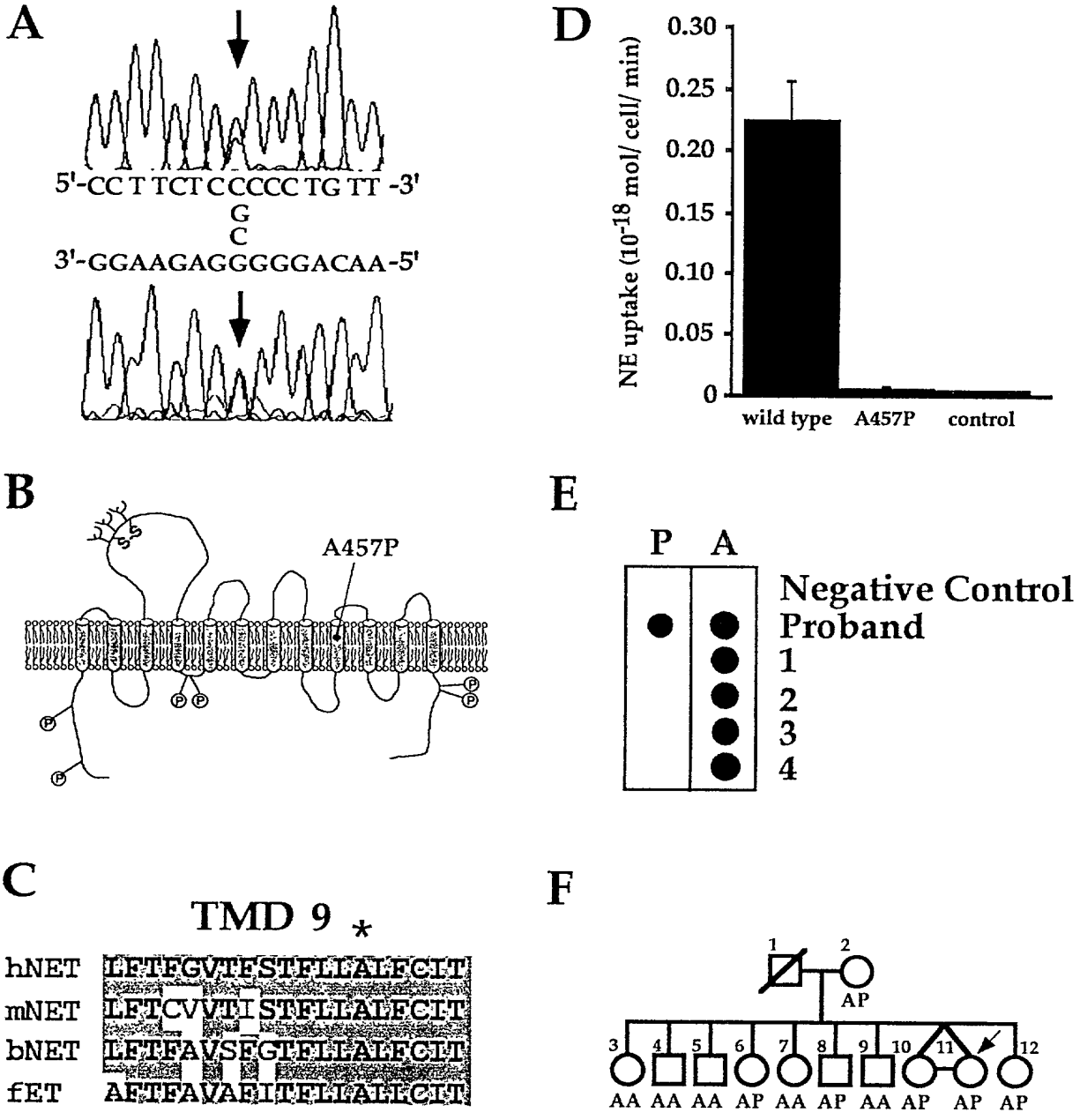

A
5'-CC T TCT C CCCC TG TT -3'
G
C
3'-GGAAGAGGGGGACAA-5'
B
A457P
C
TMD 9 *
hNET LFTFGVTFSTFLLALFCIT
mNET LFTCVVTISTFLLALFCIT
bNET LFTFAVSFGTFLLALFCIT
fET AFTFAVAFITFLLALFCIT
D
NE uptake (10-18 mol/ cell/ min)
0.30
0.25
0.20
0.15
0.10
0.05
0
wild type
A457P
control
E
P A
Negative Control
Proband
1
2
3
4
F
1 2
AP
3 4 5 6 7 8 9 10 11 12
AA AA AA AP AA AP AA AP AP AP

Figure 3

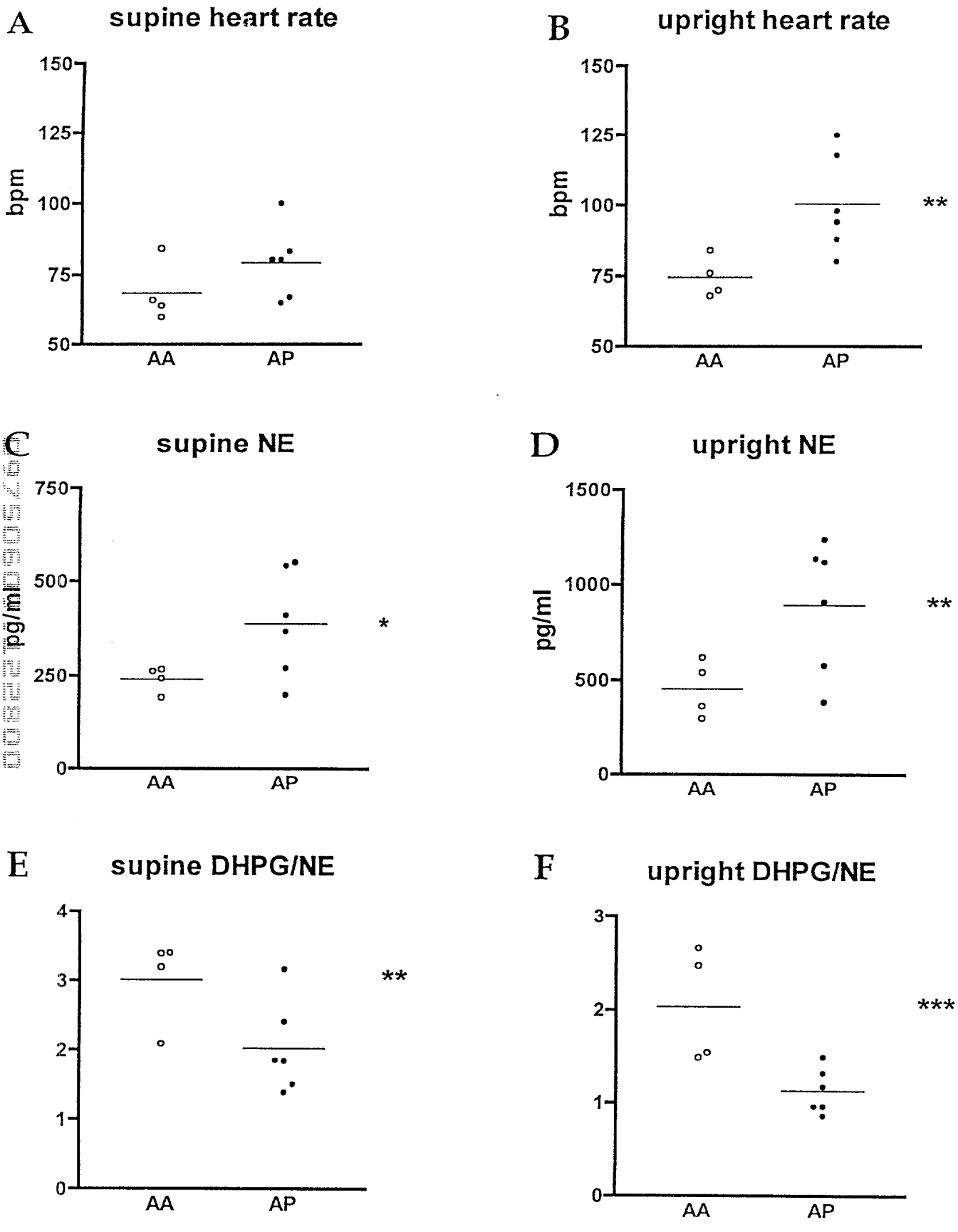
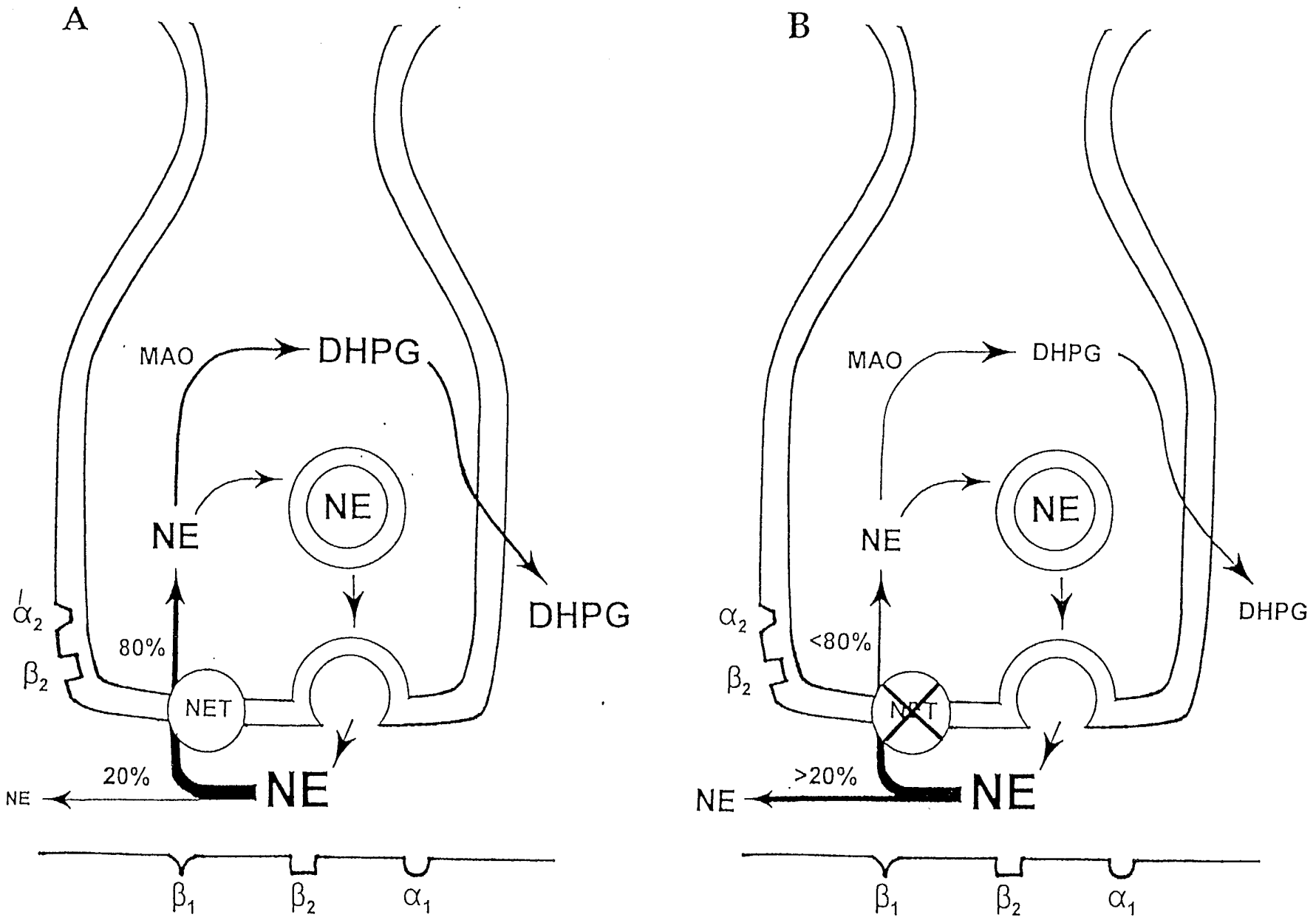

A
supine heart rate
bpm
150
125
100
75
50
AA
AP
B
upright heart rate
bpm
150
125
100
75
50
**
AA
AP
C
supine NE
pg/ml
750
500
250
0
*
AA
AP
D
upright NE
pg/ml
1500
1000
500
0
**
AA
AP
E
supine DHPG/NE
4
3
2
1
0
**
AA
AP
F
upright DHPG/NE
3
2
1
0
***
AA
AP

Figure 4


A
MAO
DHPG
NE
NE
DHPG
α_2
β_2
80%
NET
20%
NE
NE
β_1
β_2
α_1
B
MAO
DHPG
NE
NE
DHPG
α_2
β_2
<80%
NET
>20%
NE
NE
β_1
β_2
α_1